SEQUENCE	LISTING		\$110432_580	2	
<110>	Samsung El	ectronics Co.	Ltd		
<120>	A variant polymorphi	of HNF-la gene sm and a vari	e having novel ant protein enc	single nucleo oded by the s	tide ame
<160>	25				
<170>	KopatentIn	1.71			
<210> <211> <212> <213>	1 1896 DNA Homo sapie	ns			
<220> <221> <222> <223>	CDS (1)(1893 amino acid	) sequence of	HNF-1a		
<400>	1			. cta aca aca	sta 10
Met Val	tct aaa ctg Ser Lys Leu 5	ser Gln Leu	cag acg gag ctc Gln Thr Glu Leu 10	Leu Ala Ala 15	ctg 48 Leu
ctc gag Leu Glu	tca ggg ctg Ser Gly Leu 20	agc aaa gag Ser Lys Glu	gca ctg atc cag Ala Leu Ile Gln 25	gca ctg ggt Ala Leu Gly 30	gag 96 Glu
ccg ggg Pro Gly	ccc tac ctc Pro Tyr Leu 35	ctg gct gga Leu Ala Gly 40	gaa ggc ccc ctg Glu Gly Pro Leu	gac aag ggg Asp Lys Gly 45	gag 144 Glu
tcc tgc Ser Cys 50	ggc ggc ggt Gly Gly Gly	cga ggg gag Arg Gly Glu 55	ctg gct gag ctg Leu Ala Glu Leu 60	Pro Asn Gly	ctg 192 Leu
ggg gag Gly Glu 65	act cgg ggc Thr Arg Gly	tcc gag gac Ser Glu Asp 70	gag acg gac gac Glu Thr Asp Asp 75	gat ggg gaa Asp Gly Glu	gac 240 Asp 80
ttc acg Phe Thr	cca ccc atc Pro Pro Ile 85	Leu Lys Glu	ctg gag aac ctc Leu Glu Asn Leu 90	agc cct gag Ser Pro Glu 95	gag 288 Glu
gcg gcc Ala Ala	cac cag aaa His Gln Lys 100	Ala Val Val	gag acc ctt ctg Glu Thr Leu Leu 105	cag gag gac Gln Glu Asp 110	ccg 336 Pro
Trp Arg '	gtg gcg aag Val Ala Lys 115	atg gtc aag Met Val Lys 120	tcc tac ctg cag Ser Tyr Leu Gln	cag cac aac Gln His Asn 125	atc 384 Ile
			act ggc ctc aac Thr Gly Leu Asn 140	Gln Ser His	
tcc caa Ser Gln 145	cac ctc aac His Leu Asn	aag ggc act Lys Gly Thr 150	ccc atg aag acg Pro Met Lys Thr 155	cag aag cgg Gln Lys Arg	gcc 480 Ala 160

Page 1

gcc Ala	ctg Leu	tac Tyr	acc Thr	tgg Trp 165	tac Tyr	gtc val	cgc Arg	aag Lys	caq	8432. cga Arg	gag	gtg	gcg Ala	cag Gln 175	cag Gln	528
ttc Phe	acc Thr	cat His	gca Ala 180	ggg Gly	cag Gln	gga Gly	ggg Gly	ctg Leu 185	att Ile	gaa Glu	gag Glu	ccc Pro	aca Thr 190	ggt Gly	gat Asp	576
gag Glu	cta Leu	cca Pro 195	acc Thr	aag Lys	aag Lys	ggg Gly	cgg Arg 200	agg Arg	aac Asn	cgt Arg	ttc Phe	aag Lys 205	tgg Trp	ggc Gly	cca Pro	624
Ăla	tcc Ser 210	cag Gln	cag Gln	atc Ile	ctg Leu	ttc Phe 215	cag Gln	gcc Ala	tat Tyr	gag Glu	agg Arg 220	cag Gln	aag Lys	aac Asn	cct Pro	672
agc Ser 225	aag Lys	gag Glu	gag Glu	cga Arg	gag Glu 230	acg Thr	cta Leu	gtg Val	gag Glu	gag Glu 235	tgc Cys	aat Asn	agg Arg	gcg Ala	gaa Glu 240	720
tgc Cys	atc Ile	cag Gln	aga Arg	ggg G1y 245	gtg Val	tcc Ser	cca Pro	tca Ser	cag Gln 250	gca Ala	cag Gln	ggg Gly	ctg Leu	ggc Gly 255	tcc Ser	768
aac Asn	ctc Leu	gtc Val	acg Thr 260	gag Glu	gtg Val	cgt Arg	gtc Val	tac Tyr 265	aac Asn	tgg Trp	ttt Phe	gcc Ala	aac Asn 270	cgg Arg	cgc Arg	816
aaa Lys	gaa Glu	gaa Glu 275	gcc Ala	ttc Phe	cgg Arg	cac His	aag Lys 280	ctg Leu	gcc Ala	atg Met	gac Asp	acg Thr 285	tac Tyr	agc Ser	ggg Gly	864
ccc Pro	ccc Pro 290	cca Pro	ggg Gly	cca Pro	ggc Gly	ccg Pro 295	gga Gly	cct Pro	gcg Ala	ctg Leu	ccc Pro 300	gct Ala	cac His	agc Ser	tcc Ser	912
cct Pro 305	ggc Gly	ctg Leu	cct Pro	cca Pro	cct Pro 310	gcc Ala	ctc Leu	tcc Ser	ccc Pro	agt Ser 315	aag Lys	gtc Val	cac His	ggt Gly	gtg Val 320	960
cgc Arg	tat Tyr	gga Gly	cag Gln	cct Pro 325	gcg Ala	acc Thr	agt Ser	gag Glu	act Thr 330	Ala	gaa Glu	gta Val	ccc Pro	tca Ser 335	agc Ser	1008
agc Ser	ggc Gly	ggt Gly	ccc Pro 340	Leu	gtg Val	aca Thr	gtg Val	tct Ser 345	Thr	ccc Pro	ctc Leu	cac His	caa Gln 350	vaı	tcc Ser	1056
ccc Pro	acg Thr	ggc Gly 355	Leu	gag Glu	ccc Pro	agc Ser	cac His 360	Ser	ctg Leu	ctg Leu	agt Ser	aca Thr 365	GIU	gcc Ala	aag Lys	1104
ctg Leu	gtc Val 370	Ser	gca Ala	gct Ala	ggg Gly	ggc Gly 375	ccc Pro	ctc Leu	ccc Pro	cct Pro	gtc Val 380	Ser	acc T <b>h</b> r	ctg Leu	aca Thr	1152
gca Ala 385	ctg Leu	cac His	agc Ser	ttg Leu	gag Glu 390	Gln	aca Thr	tcc Ser	cca Pro	ggc Gly 395	Leu	aac Asn	cag Gln	cag Gln	ccc Pro 400	1200
cag Gln	aac Asn	ctc Leu	atc Ile	atg Met 405	Ala	tca Ser	ctt Leu	cct Pro	ggg Gly 410	' Va I	Met	acc Thr	ato	ggg Gly 415	cct Pro	1248

.

ggt Gly	gag Glu	cct Pro	gcc Ala 420	tcc Ser	ctg Leu	ggt Gly	cct Pro	acg Thr 425	ttc Phe	acc Thr	aac Asn	aca Thr	ggt Gly 430	gcc Ala	tcc Ser	1296
acc Thr	ctg Leu	gtc Val 435	atc Ile	ggc Gly	ctg Leu	gcc Ala	tcc Ser 440	acg Thr	cag Gln	gca Ala	cag Gln	agt Ser 445	gtg Val	ccg Pro	gtc Val	1344
atc Ile	aac Asn 450	agc Ser	atg Met	ggc Gly	agc Ser	agc Ser 455	ctg Leu	acc Thr	acc Thr	ctg Leu	cag Gln 460	ccc Pro	gtc Val	cag Gln	ttc Phe	1392
tcc Ser 465	cag Gln	ccg Pro	ctg Leu	сас His	ccc Pro 470	tcc Ser	tac Tyr	cag Gln	cag Gln	ccg Pro 475	ctc Leu	atg Met	cca Pro	cct Pro	gtg Val 480	1440
cag Gln	agc Ser	cat His	gtg val	acc Thr 485	cag Gln	aac Asn	ccc Pro	ttc Phe	atg Met 490	gcc Ala	acc Thr	atg Met	gct Ala	cag Gln 495	ctg Leu	1488
cag Gln	agc Ser	ccc Pro	cac His 500	gcc Ala	ctc Leu	tac Tyr	agc Ser	cac His 505	aag Lys	ccc Pro	gag Glu	gtg Val	gcc Ala 510	cag Gln	tac Tyr	1536
acc Thr	cac His	acg Thr 515	ggc Gly	ctg Leu	ctc Leu	ccg Pro	cag Gln 520	act Thr	atg Met	ctc Leu	atc Ile	acc Thr 525	gac Asp	acc Thr	acc Thr	1584
aac Asn	ctg Leu 530	agc Ser	gcc Ala	ctg Leu	gcc Ala	agc Ser 535	ctc Leu	acg Thr	ccc Pro	acc Thr	aag Lys 540	cag Gln	gtc Val	ttc Phe	acc Thr	1632
tca Ser 545	gac Asp	act Thr	gag Glu	gcc Ala	tcc ser 550	agt Ser	gag Glu	tcc Ser	ggg Gly	ctt Leu 555	cac His	acg Thr	ccg Pro	gca Ala	tct Ser 560	1680
cag Gln	gcc Ala	acc Thr	acc Thr	ctc Leu 565	cac нis	atc Ile	ccc Pro	agc Ser	cag Gln 570	gac Asp	cct Pro	gcc Ala	ggc Gly	atc Ile 575	cag Gln	1728
cac His	ctg Leu	cag Gln	ccg Pro 580	gcc Ala	cac His	cgg Arg	ctc Leu	agc ser 585	gcc Ala	agc Ser	ccc Pro	aca Thr	gtg Val 590	tcc Ser	tcc Ser	1776
agc Ser	agc Ser	ctg Leu 595	gtg Val	ctg Leu	tac Tyr	cag Gln	agc ser 600	tca Ser	gac Asp	tcc Ser	agc Ser	aat Asn 605	ggc Gly	cag Gln	agc Ser	1824
cac His	ctg Leu 610	Leu	cca Pro	tcc Ser	aac Asn	cac His 615	Ser	gtc Val	atc Ile	gag Glu	acc Thr 620	Phe	atc Ile	tcc Ser	acc Thr	1872
cag Gln 625	Met	gcc Ala	tct Ser	tcc Ser	tcc Ser 630	Gln			taa							1896
<21 <21	0> 1>	2 631														

<211> 631 <212> PRT <213> Homo sapiens

Met Val Ser Lys Leu Ser Gln Leu Gln Thr Glu Leu Leu Ala Ala Leu 1 5 10 15 Leu Glu Ser Gly Leu Ser Lys Glu Ala Leu Ile Gln Ala Leu Gly Glu 20 25 30 Pro Gly Pro Tyr Leu Leu Ala Gly Glu Gly Pro Leu Asp Lys Gly Glu 35 40 45 Ser Cys Gly Gly Gly Arg Gly Glu Leu Ala Glu Leu Pro Asn Gly Leu
50 55 60 Gly Glu Thr Arg Gly Ser Glu Asp Glu Thr Asp Asp Asp Gly Glu Asp 65 70 75 80 Phe Thr Pro Pro Ile Leu Lys Glu Leu Glu Asn Leu Ser Pro Glu Glu 85 90 95 Ala Ala His Gln Lys Ala Val Val Glu Thr Leu Leu Gln Glu Asp Pro 100 105 110 Trp Arg Val Ala Lys Met Val Lys Ser Tyr Leu Gln Gln His Asn Ile 115 120 125 Pro Gln Arg Glu Val Val Asp Thr Thr Gly Leu Asn Gln Ser His Leu 130 140 Ser Gln His Leu Asn Lys Gly Thr Pro Met Lys Thr Gln Lys Arg Ala 145 150 155 160 Ala Leu Tyr Thr Trp Tyr Val Arg Lys Gln Arg Glu Val Ala Gln Gln 165 170 175 Phe Thr His Ala Gly Gln Gly Gly Leu Ile Glu Glu Pro Thr Gly Asp 180 185 190 Glu Leu Pro Thr Lys Lys Gly Arg Arg Asn Arg Phe Lys Trp Gly Pro 195 200 205 Ala Ser Gln Gln Ile Leu Phe Gln Ala Tyr Glu Arg Gln Lys Asn Pro 210 215 220 Ser Lys Glu Glu Arg Glu Thr Leu Val Glu Glu Cys Asn Arg Ala Glu 225 230 235 240 Cys Ile Gln Arg Gly Val Ser Pro Ser Gln Ala Gln Gly Leu Gly Ser 245 250 255 Asn Leu Val Thr Glu Val Arg Val Tyr Asn Trp Phe Ala Asn Arg Arg 260 265 270 Lys Glu Glu Ala Phe Arg His Lys Leu Ala Met Asp Thr Tyr Ser Gly 275 280 285 Pro Pro Pro Gly Pro Gly Pro Ala Leu Pro Ala His Ser Ser 290 295 300 Pro Gly Leu Pro Pro Pro Ala Leu Ser Pro Ser Lys Val His Gly Val 305 310 315 320 Arg Tyr Gly Gln Pro Ala Thr Ser Glu Thr Ala Glu Val Pro Ser Ser 325 330 335 Page 4

Ser Gly Gly Pro Leu Val Thr Val Ser Thr Pro Leu His Gln Val Ser 340 345 350 Pro Thr Gly Leu Glu Pro Ser His Ser Leu Leu Ser Thr Glu Ala Lys 355 360 365 Leu Val Ser Ala Ala Gly Gly Pro Leu Pro Pro Val Ser Thr Leu Thr 370 375 380 Ala Leu His Ser Leu Glu Gln Thr Ser Pro Gly Leu Asn Gln Gln Pro 385 390 395 400 Gln Asn Leu Ile Met Ala Ser Leu Pro Gly Val Met Thr Ile Gly Pro 405 410 415 Gly Glu Pro Ala Ser Leu Gly Pro Thr Phe Thr Asn Thr Gly Ala Ser 420 425 430 Thr Leu Val Ile Gly Leu Ala Ser Thr Gln Ala Gln Ser Val Pro Val 435 440 445 Ile Asn Ser Met Gly Ser Ser Leu Thr Thr Leu Gln Pro Val Gln Phe 450 460 Ser Gln Pro Leu His Pro Ser Tyr Gln Gln Pro Leu Met Pro Pro Val 480 Gln Ser His Val Thr Gln Asn Pro Phe Met Ala Thr Met Ala Gln Leu 485 490 495 Gln Ser Pro His Ala Leu Tyr Ser His Lys Pro Glu Val Ala Gln Tyr 500 505 510 Thr His Thr Gly Leu Leu Pro Gln Thr Met Leu Ile Thr Asp Thr Thr 515 520 525 Asn Leu Ser Ala Leu Ala Ser Leu Thr Pro Thr Lys Gln Val Phe Thr 530 540 Ser Asp Thr Glu Ala Ser Ser Glu Ser Gly Leu His Thr Pro Ala Ser 545 550 555 560 Gln Ala Thr Thr Leu His Ile Pro Ser Gln Asp Pro Ala Gly Ile Gln 565 570 575 His Leu Gln Pro Ala His Arg Leu Ser Ala Ser Pro Thr Val Ser Ser 580 585 590 Ser Ser Leu Val Leu Tyr Gln Ser Ser Asp Ser Ser Asn Gly Gln Ser 595 600 605 His Leu Leu Pro Ser Asn His Ser Val Ile Glu Thr Phe Ile Ser Thr 610 615 620 Gln Met Ala Ser Ser Ser Gln 630

<sup>&</sup>lt;210> 3 <211> 93 <212> DNA

<sup>&</sup>lt;213> Homo sapiens

<400> gtaaggtc	3 ca ggcctgctgg ccctcccttg gcctgtgaca gagcccctca cccccacatc	60
ccccgggc	tc aggaggctgc tctgctcccc cag	93
<210> <211> <212> <213>	4 41 DNA Artificial Sequence	
<220> <223>	sense primer for amplifying promoter of MODY3 gene	
<400> taatacga	4 ct cactataggg tggccgtgag catcctctgc c	41
<210> <211> <212> <213>	5 39 DNA Artificial Sequence	
<220> <223>	antisense primer for amplifying promoter of MODY3 gene	
<400> gtaaccct	5 ca ctaaagggac gtgggttgcg tttgcctgc	39
<210> <211> <212> <213>	6 40 DNA Artificial Sequence	
<220> <223>	sense primer for amplifying exon 1 of MODY3 gene	
<400> taatacga	6 ct cactataggg cgtggccctg tggcagccga	40
<210> <211> <212> <213>	7 40 DNA Artificial Sequence	
<220> <223>	antisense primer for amplifying exon 1 of MODY3 gene	
<400> gtaaccct	7 ca ctaaagggag ggctcgttag gagctgaggg	40
<210> <211> <212> <213>	8 42 DNA Artificial Sequence	

<220>	of MODY2 gang	
<223>	sense primer for amplifying exon 2 of MODY3 gene	
<400> taatacga	8 act cactataggg cccttgctga gcagatcccg tc	42
<210> <211>	9 40	
<212> <213>	DNA Artificial Sequence	
<220> <223>	antisense primer for amplifying exon 2 of MODY3 gene	
<400> gtaaccct	9 cca ctaaagggag ggatggtgaa gcttccagcc	40
<210> <211>	10 40	
<212> <213>	DNA Artificial Sequence	
<220> <223>	sense primer for amplifying exon 3 of MODY3 gene	
<400> taatacga	10 act cactataggg gcaaggtcag gggaatggac	40
<210> <211>	11 42	
<212> <213>	DNA Artificial Sequence	
<220> <223>	antisense primer for amplifying exon 3 of MODY3 gene	
<400> gtaaccc	11 tca ctaaagggac gccgttgtac ctattgcact cc	42
<210> <211>	12 43	
<212> <213>	DNA Artificial Sequence	
<220> <223>	sense primer for amplifying exon 4 of MODY3 gene	
<400> taatacg	12 act cactataggg ggctcatggg tggctatttc tgc	43
<210> <211>	13 42	
. —	Page 7	

<212> <213>	DNA Artificial Sequence	
<220> <223>	antisense primer for amplifying exon 4 of MODY3 gene	
<400> gtaaccct	13 ca ctaaagggac gtgtcccttg tccccacata cc	42
<210> <211> <212> <213>	14 42 DNA Artificial Sequence	
<220> <223>	sense primer for amplifying exon 5 of MODY3 gene	
<400> taatacga	14 ct cactataggg tgctgaggca ggacactgct tc	42
<210> <211> <212> <213>	15 42 DNA Artificial Sequence	
<220> <223>	antisense primer for amplifying exon 5 of MODY3 gene	
<400> gtaaccct	15 ca ctaaagggat acaagcaagg acactcacca gc	42
<210> <211> <212> <213>	16 41 DNA Artificial Sequence	
<220> <223>	sense primer for amplifying exon 6 of MODY3 gene	
<400> taatacga	16 ct cactataggg cccggacaca gcttggcttc c	41
<210> <211> <212> <213>	17 42 DNA Artificial Sequence	
<220> <223>	antisense primer for amplifying exon 6 of MODY3 gene	
<400> gtaaccct	17 ca ctaaagggaa tccccaccag cttaccgatg ac	42

	si18432_SEQ	
<210> <211> <212>	18 40 DNA	
<213>	Artificial Sequence	
<220> <223>	sense primer for amplifying exon 7 of MODY3 gene	
<400> taatacgad	18 ct cactataggg caggcctggc ctccacgcag	40
<210> <211> <212> <213>	19 40 DNA Artificial Sequence	
<220> <223>	antisense primer for amplifying exon 7 of MODY3 gene	
<400> gtaacccto	19 ca ctaaagggag gggctctgca gctgagccat	40
<210> <211> <212> <213>	20 41 DNA Artificial Sequence	
<220> <223>	sense primer for amplifying exon 8 and 9 of MODY3 gene	
<400> taatacgad	20 ct cactataggg ggcccagtac acccacacgg g	41
<210> <211> <212> <213>	21 40 DNA Artificial Sequence	
<220> <223>	antisense primer for amplifying exon 8 and 9 of MODY3 gene	
<400> gtaacccto	21 ca ctaaagggag ggcagggaca gtaagggagg	40
<210> <211> <212> <213>	22 41 DNA Artificial Sequence	
<220> <223>	sense primer for amplifying exon 10 of MODY3 gene	

41

<400> 22 taatacgact cactataggg gccttgtttg cctctgcagt g Page 9

<210> <211> <212> <213>	23 41 DNA Artificial Sequence	
<220> <223>	antisense primer for amplifying exon 10 of MODY3 gene	
<400> gtaaccct	23 ca ctaaagggag gccatctggg tggagatgaa g	41
<210> <211> <212> <213>	24 20 DNA Artificial Sequence	
<220> <223>	T7 promoter sequence	
<400> taatacga	24 ct cactataggg	20
<210> <211> <212> <213>	25 19 DNA Artificial Sequence	
<220> <223>	T3 promoter sequence	
<400> gtaaccct	25 ca ctaaaggga	19